ABSTRACT

The present invention provides a method effective for diagnosis or treatment of congenital disorders of glycosylation syndrome (CDGS) by clarifying the gene of the *N*-linked sugar chain synthase in human endoplasmic reticulum. In the present invention, a gene of an enzyme catalyzing human *N*-linked sugar chain synthesis is found based on, as indicators, whether it is homologous with the gene of the enzyme catalyzing *N*-linked sugar chain synthesis in yeast endoplasmic reticulum and compliments the function of the gene for a deletion yeast strain of the gene.